

Patient ID 321	Patient Name TESTRNV, IMPLEMENTATION	Birth Date 1968-02-19	Sex M	Age 54
Order Number X100390258	Client Order Number X100390258	Ordering Physician UNKNOWN, PROVIDER	Report Notes	
Account Information C7028846 DLMP Rochester		Collected 30 May 2022 14:00		

Galactosemia Mutation Panel

Result Summary

MCR

POSITIVE

Result

MCR

The following homozygous variant was identified:

Amino Acid change: p.Q188R (Gln188Arg)

DNA change: c.563A>G (g.34648167)

Classification: PATHOGENIC

Interpretation

1 MCR

Biochemical and molecular test results are in agreement. The observed GALT enzyme activity in red blood cells (0.0 nmol/h/mg Hb) and the presence of two copies of Q188R (GG genotype) suggest that this individual is affected with classic galactosemia. Treatment with galactose restricted diet is recommended.

Since pathogenic variants have been identified, genetic testing of at risk family members could be considered.

A genetic consultation may be of benefit.

ADDITIONAL INFORMATION

CAUTIONS

This assay will not detect all of the known pathogenic variants causing galactosemia. Therefore, the absence of a detectable variant does not rule out the possibility that an individual is a carrier of or affected with this disease.

Many disorders may present with symptoms similar to those associated with galactosemia. Therefore, biochemical testing is recommended to establish the diagnosis of galactosemia prior to DNA analysis.

A negative result does not eliminate the risk of carrier status for any of the included conditions, due to the possibility that the patient carries a variant that is not interrogated with this assay or the rare chance of a false-negative result for a tested variant. For tested variants, the negative predictive value of this screen is

greater than 98%. The patient's residual risk to be a carrier after a negative screen is dependent on ethnic background and family history.

A positive control was not available for all variants targeted on this panel. For more information regarding availability of a positive control for each variant see Targeted Variants Interrogated by Expanded Carrier Screen Panels in Special Instructions. The negative predictive value of these targets is unknown.

Rare polymorphisms exist that could lead to false-negative or false-positive results. If results obtained do not match the clinical findings, additional testing should be considered.

All detected variants are evaluated according to American College of Medical Genetics and Genomics recommendations. This assay was designed to specifically target known pathogenic or likely pathogenic variants. In rare cases, DNA variants of undetermined significance may be identified. The laboratory encourages health care providers to contact the laboratory at any time to learn how the status of a particular variant may have changed over time.

Multiple in-silico evaluation tools may have been used to assist in the interpretation of these results. Of note, the sensitivity and specificity of these tools for the determination of pathogenicity is currently unvalidated.

Test results should be interpreted in the context of clinical findings, family history, and other laboratory data. Misinterpretation of results may occur if the information provided is inaccurate or incomplete.

Bone Marrow transplants from allogenic donors will interfere with testing. Call Mayo Clinic Laboratories for instructions for testing patients who have received a bone marrow transplant.

An online research opportunity called GenomeConnect (genomeconnect.org), a project of ClinGen, is available for the recipient of this genetic test. This patient registry collects deidentified genetic and health information to advance the

Performing Site Legend

Code	Laboratory	Address	Lab Director	CLIA Certificate
MCR	Mayo Clinic Laboratories - Rochester Main Campus	200 First Street SW, Rochester, MN 55905	William G. Morice M.D. Ph.D	24D0404292

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knowledge of genetic variants. Mayo Clinic is a collaborator of ClinGen. This may not be applicable for all tests.

Specimen

WB Whole Blood

MCR

Method

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An array based assay was used to test for the presence of the following variants in the GALT gene (GenBank accession number NM_000155; build GRCh37 (hg19)): -119_-116delGTCA, 136_140del, L74P, D98N, S135L, T138M, R148Q, M142K, Q169K, F171S, Q188R, L195P, E203K, Y209C, K285N, N314D, A320T, R333G, R333W, E340*, Q344K, 253-2A>G, *380Rext*49 and 5 kb deletion.

Released By

MCR

LINDA HASADSR

Received: 31 May 2022 16:18

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Laboratory Notes

- 1 This test was developed and its performance characteristics determined by Mayo Clinic in a manner consistent with CLIA requirements. This test has not been cleared or approved by the U.S. Food and Drug Administration.

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